Speakers

Robyn Bent, M.S., is the director of the Patient-Focused Drug Development (PFDD) Program in the Food and Drug Administration's (FDA's) Center for Drug Evaluation and Research (CDER). The PFDD is an effort to systematically obtain patient input and facilitate the incorporation of meaningful patient input into drug development and regulatory decision making. Prior to joining FDA, Captain Bent held several positions at the National Institutes of Health. She has extensive experience in clinical trial design, conduct, and oversight. She received her B.S. in nursing from The Catholic University of America and her M.S. from the George Washington University.

Philip John (P.J.) Brooks, Ph.D., is the acting director of the Division of Rare Diseases Research Innovation (DRDRI) at the National Center for Advancing Translational Sciences (NCATS). He represents NCATS on the Trans-NIH Gene Therapy Working Group and the Regenerative Medicine Innovation Project, is the Working Group Coordinator for the NIH Common Fund program on Somatic Cell Genome Editing and is one of the leaders of the PaVe-GT pilot project. He recently was elected as the chair of the Interdisciplinary Scientific Committee of the International Rare Diseases Research Consortium. Previously, Dr. Brooks was in the NCATS Division of Clinical Innovation, where he was the lead program director for the Clinical and Translational Science Awards (CTSA) Program Collaborative Innovation Awards, which are designed to fund projects that will result in novel and creative approaches to overcoming roadblocks in translational science (PAR-18-244 and PAR-18-245), and an investigator in the intramural program of the National Institute on Alcohol Abuse and Alcoholism (NIAAA). He developed an internationally recognized research program focused on two distinct areas: the molecular basis of alcohol-related cancer and rare neurologic diseases resulting from defective DNA repair, including xeroderma pigmentosum, Cockayne syndrome, and Fanconi anemia. He received his Ph.D. in neurobiology from the University of North Carolina at Chapel Hill and completed a postdoctoral fellowship at the Rockefeller University.

Chekesha Clingman-Henry, Ph.D., M.B.A., is a commander in the U.S. Public Health Service and is the associate director for Strategic Partnerships in the Office of Translational Sciences (OTS) at the Food and Drug Administration's (FDA's) Center for Drug Evaluation and Research (CDER). She advises the OTS director and senior staff on a broad range of activities to support CDER's mission, including providing strategic direction for public-private partnerships, evaluating significant regulatory science needs, and developing policies and procedures to streamline how CDER engages in scientific collaborations with external entities. In addition, Dr. Clingman-Henry is the scientific lead for the FDA Critical Path Innovation Meetings (CPIM) Program, which allows individuals from academia, nonprofit organizations, industry, and other government agencies to discuss emerging science and innovation in drug development with FDA subject matter experts in a nonbinding, nonregulatory setting. Before joining OTS, she served as a clinical analyst and regulatory reviewer within the Division of Medical Imaging Products in the CDER Office of New Drugs. She also held the position of senior consultant for strategic partnerships and intellectual property in the Office of the Chief Scientist at the FDA. Prior to joining the

FDA, she worked as a technology licensing and patent manager in the National Institutes of Health's (NIH's) Office of Technology Transfer. She received her Ph.D. in biophysics and biophysical chemistry from Johns Hopkins University and her M.B.A. from George Washington University.

Katie Donohue, M.D., is the director of the Division of Rare Diseases and Medical Genetics at the Food and Drug Administration (FDA). She joined the FDA as a medical officer in the Pulmonary and Allergy Division and then moved to the Inborn Errors Group as a clinical team lead. She received her M.D. from Virginia Commonwealth University. She completed her residency in Internal Medicine and a fellowship in Allergy and Immunology at Columbia University-New York Presbyterian Hospital. When she finished her clinical training, she joined the Columbia faculty; published original translational epidemiology research in peer-reviewed journals; and completed a master's degree in epidemiology, trial design, and biostatistics.

Sheila Farrell M.D., M.P.H., is a medical officer in the Division of Rare Diseases and Medical Genetics (DRDMG) in the Office of New Drugs (OND) at the Food and Drug Administration's (FDA's) Center for Drug Evaluation Research (CDER). Prior to joining the FDA, Dr. Farrell held several academic appointments; most recently she was an associate professor of pediatrics at the Louisiana State University Health Sciences Center (LSUHSC) in Shreveport. She received her M.D. from Georgetown University School of Medicine and completed a residency in pediatrics at Children's Hospital National Medical Center in Washington, DC. After residency she received her M.P.H. from the Johns Hopkins Bloomberg School of Public Health and completed a pharmacoepidemiology fellowship at the FDA through the Epidemiology Training Program.

Raphaela Goldbach-Mansky, M.D., M.H.S., is the chief of the Translational Autoinflammatory Diseases Section at the National Institutes of Health's (NIH's) National Institute of Allergy and Infectious Diseases (NIAID). She leads the NIAID autoinflammatory disease clinic and has built a translational research program focusing on clinical and translational studies in children with early onset autoinflammatory diseases. Together with Dr. Daniel Kastner from the National Human Genome Research Institute (NHGRI), she founded the Translational Autoinflammatory Research Initiative (TARI) at NIH to improve research in patients with rare autoinflammatory diseases. Dr. Goldbach-Mansky's research focus is on applying a systematic approach to the clinical and immunological study of autoinflammatory diseases. Her group uses targeted interventions to understand the role of specific inflammatory pathways in the pathogenesis of autoinflammatory diseases. She received her medical degree from the University Witten-Herdecke, Germany, and completed a combined residency in internal medicine and pediatrics at Case Western Reserve University's Metro Health Medical Center. She completed her rheumatology fellowship training at the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS) in 1999 and served as a staff clinician at NIAMS through 2008.

Leslie Gordon, M.D., Ph.D., is a co-founder and the medical director of The Progeria Research Foundation (PRF) and is the principal investigator for ongoing PRF programs for progeria, including an International Patient Registry, Medical and Research Database, Cell and TissueBank, and Diagnostics Program. She also is a professor of pediatrics at Hasbro Children's Hospital and Alpert Medical School of Brown University in Providence, Rhode Island; a research associate in anesthesia at Harvard Medical School and Boston Children's Hospital; and a research scientist at Women & Infants Hospital in Boston, Massachusetts. She has organized eight National Institutes of Health-funded international scientific meetings on progeria. Dr. Gordon was co-author on the 2003 gene discovery for progeria, lead author of the 2012 Progeria Treatment Discovery Study, and is co-chair of four progeria clinical drug trials at Boston Children's Hospital. She has received the March of Dimes Basil O'Connor Award, the American Heart Association Scientist Development Award, The Gerontological Society of America Award for contributions to progeria, a National Institutes of Health Bench to Bedside Grant, and the Mother of the Year award from *Working Mother Magazine*. She earned her bachelor's degree in zoology from the University of New Hampshire and her master's degree and medical and doctorate degrees from Brown University and the Brown University School of Medicine, respectively, where she achieved top honors in the medical program.

Andrea Gropman, M.D., is a professor of pediatrics, neurology, genomics, and personalized medicine at George Washington University; the division chief for Neurogenetics and Neurodevelopmental Disabilities; and the principal investigator (PI) for the Urea Cycle Rare Disorders Consortium. She also serves leadership roles on the Rare Diseases Clinical Research Network (RDCRN) and in genetic, metabolic, and neurology societies. She has published more than 200 articles and reviews; contributed chapters to classic textbooks in genetics and neurology; is one of the editors of the classic textbook, *Swaiman's Pediatric Neurology*; and is coauthor of the textbook, *X and Y Chromosome Variations*. Dr. Gropman's research has focused on several areas including inborn errors of metabolism, in which she is considered an international authority on neuroimaging and brain biomarkers in urea cycle disorders; mitochondrial epigenomics with long-time collaborator Dr. Anne Chiaramello; and chromosome disorders including Smith-Magenis syndrome and X and Y chromosome disorders. She has dedicated her career to the care of children and adults with rare disease. She is board certified in neurology/child neurology, genetics, biochemical genetics, and neurodevelopmental disabilities. She received her M.D. from the University of Massachusetts School of Medicine and completed a residency in pediatrics at Johns Hopkins Hospital. She completed subsequent fellowships in neurology/child neurology at George Washington University and Children's National Hospital; clinical and biochemical genetics at the National Institutes of Health; and a mini fellowship in neuroimaging and magnetic resonance spectroscopy at the Huntington Medical Research Institute.

Margaret Kober, R.Ph., M.P.A., is a chief of the Project Management Staff in the Office of Regulatory Operations within the Office of New Drugs at the Food and Drug Administration's (FDA's) Center for Drug Evaluation and Research (CDER). In this capacity, she provides supervisory leadership and direction to the project management staff; participates in the development and implementation of policy and strategy for the regulation of reproductive, urologic, dermatologic, and dental product classes; and participates in meetings with industry to provide information and advice on policy, law, and regulations. Previously, Ms. Kober held the position of regulatory review officer in the Division of Drug Marketing, Advertising, and Communications (DDMAC) in CDER. She was responsible for the regulation of promotional activities related to metabolic and endocrine drug products. She also has served as a team leader for the Drug Registration and Listing Project. Prior to joining FDA, she had 15 years of experience in community pharmacy practice. She received her B.S. in pharmacy from the University of Rhode Island and her M.P.A. with a concentration in health policy and administration from George Mason University.

Matthias Kretzler, M.D., is the Warner-Lambert/Parke-Davis Professor of Internal Medicine/Nephrology and Computational Bioinformatics at the University of Michigan. The overarching goal of his research is to define rare glomerular diseases in mechanistic terms and use this knowledge for targeted therapeutic interventions. To reach this goal his international research team at the University of Michigan has developed a translational research pipeline centered on integrated systems biology analysis of renal disease. Over the last 25 years the team has built experience in interdisciplinary data integration of large-scale data sets in international multi-disciplinary research networks and public-private partnerships across diseases and continents. The networks Dr. Kretzler helps to coordinate (NEPTUNE, KPMP, CPROBE and RPC2) link carefully monitored environmental exposures, genetic predispositions, transcriptional networks, proteomic profiles, metabolic fingerprints, digital histological biopsy archives, and prospective clinical disease characterizations to define cross-cutting disease mechanisms. The molecular mechanisms identified have resulted in more than 350 publications and, most importantly, in new disease predictors, de novo drug development, and successful clinical trials of novel therapeutic modalities for glomerular diseases. He received his M.D. from Heidelberg University.

Brendan Lee, M.D., Ph.D., is the Robert and Janice McNair Endowed Chair in Molecular and Human Genetics and a professor and chairman of the Department of Molecular and Human Genetics at Baylor College of Medicine (BCM). As a pediatrician and geneticist, he studies structural birth defects and inborn errors of metabolism. He currently leads the National Institutes of Health (NIH) BCM Undiagnosed Diseases Network Clinical Site at BCM and the NIH Brittle Bone Disorders Consortium. He holds multiple patents in drug discovery and gene therapy and several licensed technologies are in clinical trial including in osteogenesis imperfecta, osteoarthritis, and maple syrup urine disease (MSUD). He also has led the development of clinical studies that led to Food and Drug Administration (FDA)-approved therapies in urea cycle disorders. Dr. Lee has received local, national, and international recognition including election to the National Academy of Medicine and Fellow of the American Association for the Advancement of Science (AAAS) and membership in the Association of American Physicians (AAP), the American Society for Clinical Investigation (ASCI), and the Society of Pediatric Research (SPR). He has been awarded the ASHG Curt Stern Award for Outstanding Scientific Achievement, the SPR E. Mead Johnson Award for Pediatrics Research, and the American Philosophical Society's Judson Darland Prize for Patient-Oriented Clinical Investigation. He received his M.D. and Ph.D. from the State University Of New York Downstate Medical Center.

Kerry Jo Lee, M.D., is the associate director for rare diseases in the Office of New Drugs (OND) at the Food and Drug Administration's (FDA's) Center for Drug Evaluation and Research (CDER). She is a physician-scientist with a specialty in pediatric disease of the gastrointestinal and hepatic systems with a special focus on the microbiome and its interplay in health and disease in these areas. Throughout her career she has maintained a steadfast interest in policy and ethics and has continued to serve on committees for both. Previously she was a clinical advisor for OND policy at CDER and a medical officer in the Division of Gastroenterology and Inborn Errors of Metabolism. Dr. Lee worked with the National Bioethics Advisory Commission on reports that advised on ethical and policy issues in international and domestic clinical trials and interned at the World Health Organization. She received her B.A. in ecology and evolutionary biology from Princeton University and her M.D., with an honors degree conferred in microbiology, from the New York University School of Medicine. She completed a residency in pediatrics at the Children's Hospital of Los Angeles and a postdoctoral clinical fellowship in pediatric gastroenterology, hepatology, and nutrition at Columbia University College of Physicians and Surgeons.

Janet Maynard, M.D., M.H.S., is the director of the Office of Rare Diseases, Pediatrics, Urologic and Reproductive Medicine (ORPURM) at the Food and Drug Administration (FDA). She oversees the development, review, and regulation of applications for drugs and biologic products reviewed in the divisions in ORPURM: the Division of Pediatrics and Maternal Health (DPMH); the Division of Rare Diseases and Medical Genetics (DRDMG); the Division of Urology, Obstetrics, and Gynecology (DUOG); and the Division of Pharmacology-Toxicology for Rare Diseases, Pediatrics, Urologic and Reproductive Medicine/Specialty Medicine (DPT-RPURM/SM). Prior to serving as the director, she was the deputy director of ORPURM. Previously Dr. Maynard was the director of the Office of Orphan Products Development (OOPD) and oversaw the legislatively mandated designation and grant programs intended to promote the development of products for rare diseases, including orphan drug, rare pediatric disease, and humanitarian use device designation programs as well as clinical trial, natural history study, and pediatric device consortia grant programs. Prior to OOPD, she worked in the Center for Drug Evaluation and Research (CDER), where she was a clinical team leader in the Division of Anesthesia, Analgesia, and Addiction Products (DAAAP). Dr. Maynard has been with FDA since 2011, when she joined FDA's Division of Pulmonary, Allergy, and Rheumatology Products (DPARP) as a medical officer before becoming a clinical team leader in DPARP. She received her M.D. from Vanderbilt University and completed a residency in internal medicine at Duke Hospital. She then completed a fellowship in rheumatology at Johns Hopkins Hospital and, during her fellowship, completed her M.H.S. at the Johns Hopkins Bloomberg School of Public Health in the Graduate Training Program in Clinical Investigation.

Arianne Motter, Ph.D., is a board-certified senior toxicologist in the Division of Pharmacology and Toxicology for Infective Diseases at the U.S. Food and Drug Administration (FDA), where she reviews nonclinical studies for antiviral drug products. She also is an adjunct assistant professor in the Department of Pharmacology & Physiology at Georgetown University. Dr. Motter has been with the FDA for 8 years and actively works on investigational new drug (IND), emergency use authorization (EUA), new drug application (NDA), and biologics license application (BLA) submissions. Prior to the FDA, she was a toxicologist with the Armed Forces Medical Examiner. She received her Ph.D. in pharmacology from Georgetown University.

Elizabeth Ottinger, Ph.D., is a senior program manager and drug development team lead in the Therapeutics for Rare and Neglected Diseases (TRND) Program in the Division of Preclinical Innovation at the National Institutes of Health's (NIH's) National Center for Advancing Translational Sciences (NCATS). In this role, she manages programs with the objective of developing therapies for rare and neglected diseases. She manages project teams, developing and implementing project plans to advance drug candidates through all phases of development, from preclinical to clinical testing. Dr. Ottinger proactively builds collaborations, fostering productive team environments in which the many stakeholders—including TRND leadership, technology transfer officers, collaborators, and contract research organizations—work closely to drive projects forward to a successful outcome. She has experience across multiple therapeutic modalities, including small molecules, peptides, and biologics. She has worked on treatments for neurological and metabolic diseases, cancer, lysosomal storage disorders, and infectious diseases. Dr. Ottinger has extensive experience in drug discovery, both in the pharmaceutical industry and in academic research. Prior to joining NCATS in 2010, she was a research fellow in the vaccine department at Merck Research Laboratories, where she worked on developing a broad range of bacterial and anti-viral vaccines. For part of her career, she was the manager of a high-throughput screening laboratory at the University of Pennsylvania, where she developed biochemical and cell-based assays for high-throughput screening of targets for spinal muscular atrophy. She also was an assistant professor of chemistry at Kenyon College and Swarthmore College, where she focused on teaching, research, and mentoring undergraduate students. She received her undergraduate degree in chemistry from Franklin and Marshall College and her Ph.D. in chemistry from the University of Minnesota. She completed her postdoctoral training at the Joslin Diabetes Center of Harvard Medical School.

Jennifer Rodriguez Pippins, M.D., M.P.H., is a clinical advisor in the Office of New Drug Policy (ONDP) in the Office of New Drugs (OND) at the Food and Drug Administration's (FDA's) Center for Drug Evaluation and Research (CDER). She leads the development of policy-related guidance, provides internal training on policy-related matters, and advises OND and CDER stakeholders on clinical and regulatory policy issues. Prior to ONDP, she served as an acting associate director for Clinical Programs in the Office of Medical Products and Tobacco (OMPT) in the Office of the Commissioner. She provided clinical expertise to inform OMPT's cross-center initiatives, including work on youth tobacco cessation and orphan product development. Prior to OMPT, Dr. Pippins was the deputy division director for safety in the Division of Metabolism and Endocrinology Products (DMEP) in the OND, where she oversaw the division's work on risk evaluation and management strategies (REMS) both pre- and post-approval, the evaluation of emerging safety signals, the approval of safety labeling changes, and the issuing of drug safety communications. Prior to DMEP, she was a medical officer in the Division of Pulmonary, Allergy, and Rheumatology Products (DPARP). She received her M.D. from Harvard Medical School and completed a residency at the Harvard Combined Med/Peds Program. Subsequently, she completed a general internal medicine research fellowship at Brigham and Women's Hospital, during which she completed her M.P.H. with a concentration in clinical effectiveness at the Harvard School of Public Health.

Bita Shakoory, M.D., is the clinical research manager of the Translational Autoinflammatory Diseases Section at the National Institutes of Health's (NIH's) National Institute of Allergy and Infectious Diseases (NIAID). She is an experienced clinical researcher with a demonstrated history of working in both academia as a skilled rheumatologist and in the research industry as a medical director of medical affairs. Previous positions include visiting researcher at the NIH, director of medical affairs at PRA Health Sciences, assistant professor of medicine at George Washington University, assistant professor of medicine at Temple University, and researcher and assistant professor of medicine at the University of Alabama at Birmingham. She received her M.D. from Zanjan University of Medical Sciences.

Jeffrey Siegel, M.D., is the director of the Office of Drug Evaluation Sciences (ODES) in the Office of New Drugs (OND), at the Food and Drug Administration's (FDA's) Center for Drug Evaluation and Research (CDER). ODES oversees clinical outcome assessments, biomarker qualification, research, and bioinformatics in OND. Dr. Siegel has more than 20 years of experience in research, regulatory, and clinical drug development. He served at FDA as a medical officer and then medical team leader. He left FDA for industry and worked at Genentech/Roche as global lead for rheumatology and rare diseases and then at Gilead Sciences as translational medicine lead in clinical research/inflammation before rejoining FDA. He received his B.A. from Columbia University and his M.D. from Yale University, trained in internal medicine at the University Hospitals of Cleveland, and completed a fellowship in immunology and signal transduction at the National Institutes of Health (NIH).

Mari Suzuki, M.D., is a medical officer in the Office of New Drugs at the Food and Drug Administration's Center for Drug Evaluation and Research, where she reviews and offers advice on investigational, new biologic, and new drug applications in rare diseases. She received her medical degree from the George Washington University School of Medicine and Health Sciences and completed an internal medicine residency at New York-Presbyterian Hospital before completing an inter-institute endocrinology fellowship at the National Institutes of Health (NIH). While at NIH, she was a rare disease investigator.

Shamir Tuchman, M.D., M.P.H., is a medical officer in the Division of Pediatrics and Maternal Health (DPMH) at the Food and Drug Administration (FDA). He works within the DPMH providing consultation to review divisions for varied topics relating to drug product and device development for pediatric patients. Prior to joining the FDA, he was an academic pediatric nephrologist in the Division of Pediatric Nephrology at Children's National Hospital and an associate professor of pediatrics at The George Washington University School of Medicine. His research and clinical focus areas during my career in academic medicine were on bone and mineral metabolism abnormalities in pediatric patients with chronic kidney disease. He also was the Pediatric Nephrology Fellowship Program Director at Children's National Hospital.

Tiina Urv, Ph.D., is the program director for the Rare Diseases Clinical Research Network (RDCRN), a multidisciplinary international program in the National Center for Advancing Translational Sciences (NCATS) Division of Rare Diseases Research Innovation (DRDRI). As the lead for the RDCRN program, she collaborates with 10 National Institutes of Health (NIH) Institutes to manage 22 consortia and a central Data Management Coordinating Center. The RDCRN has more than 200 participating sites in 17 countries and more than 100 Patient Advocacy Groups as research partners and conducts research on about 200 rare diseases. Before joining the DRDRI, she was a program director in the Division of Clinical Innovation, where she provided stewardship for multiple Clinical and Translational Science Awards Program hubs and worked with the Trial Innovation Network as well as with NCATS's DRDRI. Previously she worked as a program director in the Intellectual and Developmental Disabilities Branch at the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD), where she coordinated the Hunter Kelly Newborn Screening Research Program; chaired the trans-NIH Fragile X research program; and managed a diverse portfolio of basic, behavioral, and biobehavioral research related to developmental disabilities and rare diseases. Prior to joining NIH, she was an assistant professor at the University of Massachusetts Medical School's Eunice Kennedy Shriver Center and a research scientist at the New York State Institute for Basic Research in Developmental Disabilities where her research focused on Alzheimer's disease in individuals with Down syndrome. She received her B.A. in sociology from the University of Washington and her M.A. in applied behavior analysis and her Ph.D. in intellectual and developmental disabilities from Columbia University.

Jie (Jack) Wang, Ph.D., is the team leader for the Rare Diseases and Inborn Errors of Metabolism Review Team in the Division of Translational and Precision Medicine (DTPM) in the Food and Drug Administration's (FDA's) Office of Clinical Pharmacology (OCP). He also currently serves as the vice chair of the Biologics Oversight Board in the OCP, a steering committee member of the Rare Disease Scientific Interest Group in the OCP, and a member of the Immunogenicity Working Group in the Center for Drug Evaluation and Research (CDER). Dr. Wang joined the FDA in 2011 and has served as a reviewer and team leader for clinical pharmacology review teams responsible for evaluating investigational new drugs (INDs), new drug applications (NDAs), and biologics license applications (BLAs) for drugs and biologics in therapeutic areas including dermatology, dentistry, urology, obstetrics, gynecology, gastroenterology, and inborn errors of metabolism. He has contributed to drafting multiple policy briefs and guidance at the FDA. He has served as principal investigator of multiple research projects funded by the National Institutes of Health (NIH) and FDA and has authored or co-authored 30 peer-reviewed journal articles and 40 abstracts in the areas of pharmacokinetics, pharmacodynamics, biopharmaceutics, immunogenicity, gene therapy, and nanomedicine. He received his B.S. in pharmacy from Beijing Medical University, his M.S. in pharmaceutical sciences from Peking University, and his Ph.D. in pharmaceutics from The Ohio State University.

Yan Wang, Ph.D., is a statistical team leader in the Division of Biometrics IV in the Office of Biostatics at the Food and Drug Administration's (FDA's) Center for Drug Evaluation and Research (CDER), providing statistical leadership and support to the Division of Rare Disease and Medical Genetics. Dr. Wang has been at FDA for more than 15 years and has played an active role in the development and application of statistical methodology used in the regulation of a variety of therapeutic areas including anti-infective, ophthalmology, transplant, and rare diseases. She received her Ph.D. in biostatistics from the University of California, Los Angeles.

Cynthia Welsh, M.D., is a medical officer on the Rare Diseases Team in the Division of Rare Diseases and Medical Genetics at the Food and Drug Administration's (FDA's) Center for Drug Evaluation and Research (CDER). Dr. Welsh joined the FDA more than 15 years ago, initially as a reviewer of diagnostic and therapeutic radiopharmaceuticals and radiation medical countermeasures prior to moving to the Rare Diseases Team. She received her B.S. from the University of California, Irvine, and her M.D. from Georgetown University and completed her residency in radiation oncology at Georgetown University.